Public Workshop
Complex Issues in Developing Drug and Biological Products for Rare Diseases
January 7, 2014

Speakers' and Panelists' Biographies

**Alphabetically ordered by last name*

PETER ADAMSON, M.D., Children's Oncology Group

Peter C. Adamson, M.D. is Chair of the Children's Oncology Group (COG), a National Cancer Institute (NCI) supported international consortium of more than 220 childhood centers that conducts clinical-translational research, including large-scale clinical trials, in children with cancer. He is Professor of Pediatrics and Pharmacology at the University of Pennsylvania School of Medicine and Chief of the Division of Clinical Pharmacology and Therapeutics at The Children's Hospital of Philadelphia. Dr. Adamson is Board Certified in Pediatric Hematology/Oncology and in Clinical Pharmacology. He is an internationally recognized leader in pediatric cancer drug development, having served until 2008 as Chair of the COG's Developmental Therapeutics Program. Prior to becoming Chair of the COG, Dr. Adamson served as Director for Clinical and Translational Research at The Children's Hospital of Philadelphia. His laboratory focuses on the clinical pharmacology of new drugs for childhood cancer.

DAVID ARONS, J.D., National Brain Tumor Society

David Arons is Senior Director of Public Policy at National Brain Tumor Society, the largest nonprofit organization in the U.S. dedicated to the brain tumor community with a mission to find new and effective therapies and ultimately a cure to brain tumors. Mr. Arons directs the organization's public policy and advocacy program focusing on advancing scientific research and improving health care for brain tumor patients, caregivers and families. He is a graduate of William Mitchell College of Law and has a master's degree in public policy from Tufts University.

ROBERT BEALL, Ph.D., Cystic Fibrosis Foundation

Under the leadership of Robert J. Beall, Ph.D., the Cystic Fibrosis Foundation has significantly advanced the search for a cure for cystic fibrosis and helped dramatically improve the length and quality of life for people with CF. Dr. Beall began his tenure at the CF Foundation in 1980 as Executive Vice President for Medical Affairs and for the past 19 years has served as President and Chief Executive Officer. Prior to joining the CF Foundation, Dr. Beall was on the medical school faculty of Case Western Reserve University in Cleveland, and worked at the National Institutes of Health, where he managed a large portion of NIH's cystic fibrosis program.

During Dr. Beall's tenure, the CF Foundation has become one of the most respected voluntary health organizations in the country. The CF Foundation's innovative "venture philanthropy" approach to drug development, in which the CF Foundation invests in research collaborations with pharmaceutical companies to help bring new therapies to patients, has been emulated by other nonprofit health organizations. The creation in the 1980s of a national network of research centers of excellence, the Research Development Program, attracted many leading institutions and first-rate scientists to the CF research effort. This collaborative network contributed to the identification of the CF gene in 1989. Since that discovery, the CF Foundation has focused on translating knowledge about the gene and the basic defect in CF into the development of novel CF treatments. In 1998, the CF Foundation launched its ground-breaking Therapeutics Development Program, a unique coalition of industry, academics and the CF Foundation that is directed at speeding the discovery and development of cutting-edge new CF drugs.

Nearly every CF drug available today was made possible because of the CF Foundation's business model, most recently Kalydeco, the first drug to treat the underlying cause of the disease in a segment of the CF population. The CF Foundation maintains a robust pipeline of potential CF therapies that target the disease from every angle, and the prospects for a cure for cystic fibrosis have never been higher.

DANIEL BENJAMIN JR., M.D., M.P.H., Ph.D., Duke University of School of Medicine

Dr. Benjamin obtained his medical degree and completed a residency in pediatrics at the University of Virginia in 1995 and 1998, respectively. He completed a fellowship in pediatric infectious disease at Duke University, obtained a MPH at the University of North Carolina (UNC) Chapel Hill, and accepted a faculty appointment at Duke University in 2001. He completed a PhD in Epidemiology at the UNC Chapel Hill in 2003. He is the author of over 180 peer-reviewed publications, most of which are in neonatal infectious disease and therapeutics.

He was the principal investigator and protocol chair of a multi-center trial for a new antimicrobial agent tested for the first time in children. Those data were submitted to FDA and included in the FDA label. Dr. Benjamin wrote the protocol, recruited the sites, and is the lead author of the published manuscript for that trial. He was the principal investigator and protocol chair of a 25-site blinded randomized trial for a new polyclonal antibody to prevent sepsis in VLBW infants. Those data were submitted to FDA. Dr. Benjamin wrote the protocol, recruited the sites, and is the lead author of the published manuscript for that trial. He is the principal investigator and protocol chair of a trial in 1,500 ELBW infants that is a public-private partnership between NICHD and industry. Dr. Benjamin wrote that protocol. He has been the Protocol Chair for five multi-center trials investigating the PK of antimicrobial in neonates and young children. He wrote each of those protocols.

Dr. Benjamin is Professor of Pediatrics and Associate Director of the Duke Clinical Research Institute. He has won 7 teaching and mentorship awards as a pediatric resident, subspecialty fellow, and faculty member. He was the first Director of Duke Clinical Research Fellowship Program, a program that now has over 30 postdoctoral fellows and medical students. He has

secured extramural funding for mentorship (1K24HD058735), and is the Duke PI for joint fellowship training program in clinical pharmacology with UNC supported by the NIH (1T32GM086330) and industry. The program has 9 active fellows in obstetrics, pediatrics, geriatrics, and PhD-trained pharmacologists. Dr. Benjamin has mentored over two-dozen college students, medical students, residents, fellows and junior faculty; 40% of his mentees are female and 25% are URM. He has 137 peer-reviewed publications with trainees as first author. Four current faculty mentees have NIH support as a PI including K23 and R18 funding; two are now Associate Professors. Three additional faculty mentees have NIH funding undergoing peer review. Dr. Benjamin has served as either chair or committee member for 16 Master's theses or PhD dissertations; of these trainees, 75% are female and 31% are members of URM groups. Dr. Benjamin is Chair and Principal Investigator for the Pediatric Trials Network, a $95 million NIH-sponsored clinical trials and pharmaco-epidemiology initiative that also functions as a platform to train junior faculty and fellows in clinical research. The Network has 20 active studies, 19 of which are led by a junior investigator, under the mentorship of Dr. Benjamin.

Dr. Benjamin held a joint appointment at the Food and Drug Administration in the Office of the Commissioner. In that role he provided advice to FDA in pediatric and neonatal drug development across all ages and therapeutic areas. His work with FDA includes an analysis of the utility of non-inferiority trial design, dose-response in antihypertensive agents, the economics of Pediatric Exclusivity (Li et al JAMA 2007), dissemination of data from the Exclusivity Program (Benjamin et al JAMA 2006), and the use of animal models in the process of drug development in neonatal candidiasis. His thinking on non-inferiority trial design has subsequently impacted all non-inferiority trials in pediatric infectious disease for labeling; his work in antihypertensive trial design has changed dose escalation strategies for trials submitted for labeling to FDA and to the European Medicines Agency (EMEA): and his peer-reviewed publications on the success of Exclusivity was quoted and used in Senate deliberations on the recent renewal of the Best Pharmaceuticals for Children Act.

As proof of concept of his ability to secure funding for trainees, Dr. Benjamin has secured the tuition for 17 trainees to obtain masters or PhD training over the past 3 years from extramural sponsorship.

AMY CELENTO, Cooley's Anemia Foundation

Amy Celento serves as the Patient Representative on the FDA's Pediatric Advisory Committee.

Amy is actively involved in a volunteer professional capacity, and a personal level, with the thalassemia national advocacy organization - The Cooley's Anemia Foundation, Inc. as the National Vice President, and founder and leader of the North Jersey Chapter.

Amy speaks on thalassemia screening and treatment to various social and service groups; writes occasional articles distributed through the Cooley's Anemia Foundation; facilitates parent-to-parent psychosocial session at Thalassemia Action Group conference; and counsels new parents with children who have been diagnosed with thalassemia.

Amy worked in marketing, account and product management, and retail administration in banking and global payment systems for 22 years.

KATIE CLAPP, M.S., FRAXA Research Foundation

Katie Clapp is president and executive director of FRAXA Research Foundation. In 1994, after her son Andrew was diagnosed with Fragile X syndrome, Katie and other parents launched FRAXA to accelerate research aimed at finding specific treatments and a cure for Fragile X. Katie has a BA in History from Harvard University and a Master degree in Computer Science from the University of North Carolina at Chapel Hill. She worked as a software engineer and adjunct professor in computer science before founding FRAXA. She has won a number of prizes for her work with FRAXA, including a 2003 Community Hero award from the American Red Cross and a 2008 Women of Justice award from Massachusetts Lawyers Weekly.

LAWRENCE CHARNAS, M.D., Ph.D., Shire Pharmaceuticals

Lawrence Charnas, MD, PhD is currently Director & Head of Discovery Medicine, Research at Shire Pharmaceuticals in Lexington, Massachusetts. He joined Shire in 2008 as a Medical Director, Translational Medicine in the legacy HGT Division, where he was the Medical lead on several clinical development programs including ACE031, a soluble ActRIIb receptor for Duchenne Muscular Dystrophy and an enzyme replacement therapy in Globoid Cell Leukodystrophy (Krabbe Disease). Prior to joining Shire, he was an Associate Professor of Pediatrics and Neurology at University of Minnesota, working on the treatment of Adrenoleukodystrophy and other lysosomal leukodystrophies with Hematopoietic Cell Transplant. Prior to joining the faculty at University of Minnesota, he had developed expertise in a number of other rare, pediatric genetic diseases including cystinosis, osteogenesis imperfecta and the Oculocerebrorenal syndrome of Lowe. He received his B.A. in Chemistry (Cornell University, Ithaca, NY), an MD and PhD - Genetics (University of Pennsylvania, Philadelphia, PA) and clinical training in Adult Neurology (Johns Hopkins Hospital, Baltimore, MD), Medical and Biochemical Genetics (NICHD, Bethesda, MD), and Pediatric Neurology (University of Minnesota). He has co-authored many peer reviewed articles, and serves on the Medical and Scientific Advisory Boards of the MLD Foundation and Lowe Syndrome Association.

EDWARD CONNOR, M.D., M.B.E., Children's National Health System & Clinical and Translational Sciences Institute at Children's National

Dr. Connor currently is Director of the Office of Innovation Development at Children's National Medical Center; Professor of Pediatrics, Microbiology, Immunology, and Tropical Medicine at George Washington School of Medicine and Health Sciences; Co-Director, Innovative Strategies and Services (ISS), Clinical and Translational Research Institute at Children's National (CTSI-CN) in Washington, DC. He attended the University of Pennsylvania School of Medicine (MD 1987 and Masters in Bioethics 2012); and did postgraduate training as a resident and chief

resident in Pediatrics at Northwestern University School of Medicine, Children's Memorial Hospital); and as a fellow in Pediatric Infectious Diseases at the University of Rochester School of Medicine and Dentistry. Dr. Connor has more than 30 years experience in clinical and translational research and product development in academia and biotechnology. This included serving as PI of the Pediatric AIDS Clinical Trials Unit at New Jersey Medical School, Chairman of the national NIAID-ACTG Pediatric Committee, and PI of ACTG 076 that led to FDA approval of zidovudine for prevention of mother-to-infant transmission of HIV. During that time, Dr. Connor served as a consultant and advisor to NIH, FDA, CDC, WHO and other authorities and led efforts to establish standards of care for HIV infected children and pregnant women. Dr. Connor subsequently moved to the biotechnology industry, where he was the Executive Vice President and Chief Medical Officer at MedImmune. There he directed all of the company's clinical development programs, including ones leading to regulatory approval of RespiGam™ and Synagis™ for prevention of serious RSV and the live attenuated intranasal influenza vaccine (FluMist™). Dr. Connor also was responsible for Medical Affairs, Product Safety, and Clinical Operations, including Biostatistics/Data Management and was Co-Chair of the company's product development committee.

Dr. Connor joined Children's National Medical Center in 2008. The Office of Innovation Development at Children's National is involved with management of intellectual property, technology transfer, partnerships, and product development (drugs, devices, and others). This includes educational programs in academic entrepreneurship and innovation, policy, regulatory, and ethics. Dr. Connor serves as the Director of the LEAP Fellowship in Pediatric Academic Entrepreneurship, provides mentorship and direction to investigators in product development and Clinical and Translational Research, and serves as senior faculty at Children's Research Institute. Dr. Connor also serves as CEO and Chief Medical Officer of ReveraGen (a company created as part of technology transfer at Children's National) and as a consultant in pediatric product development in public, academic and private sectors. He is the PI of a Department of Defense grant for drug development in Duchenne Muscular Dystrophy and investigator in an NIH U54 Award in Developmental Pharmacology.

Dr. Connor also serves in leadership roles in Clinical and Translational Science Institute at Children's National, a partnership with George Washington University and part of the Clinical and Translational Science Award (CTSA) Consortium funded by the National Center for Advancing Translational Science, NIH. Dr. Connor is a member of the Executive Committee of the CTSI-CN, Co-Director of the Pediatrics Therapeutics Development and Chair-Elect of the national CTSA'S Child Health Oversight Committee.

ALEJANDRO DORENBAUM, M.D., Lumena Pharmaceuticals

Dr. Dorenbaum received his training in medicine at The National Autonomous University in Mexico City, and Pediatric and Allergy Immunology training at the University of Texas Health Science Center and at Baylor College of Medicine, respectively. Dr. Dorenbaum joined the faculty of the University of California in San Francisco where he founded the Adolescent HIV

program and the Women and Children's Specialty Service, a program devoted to the care of women and children with HIV infection. He was recipient of two coveted awards in teaching.

Dr. Dorenbaum joined Chiron Corporation where he acquired broad expertise in several areas of drug development including biologics, small molecules, and vaccines. Dr. Dorenbaum also worked at BioMarin Pharmaceutical, Inc. where he conducted the clinical development of Kuvan®, the first drug approved for the management of patients with Phenylketonuria and Genentech, where he was responsible for the respiratory programs for asthma and cystic fibrosis. Currently, Dr. Dorenbaum is the Chief Medical Officer at Lumena Pharmaceutical. He maintains an active academic position as Associate Clinical Professor of Pediatrics at Stanford University School of Medicine, were he treats patients with Allergy and Immunology clinical problems.

ANNE-VIRGINIE EGGIMANN, M.Sc., bluebird bio, Inc.

Anne-Virginie joined bluebird bio, a start-up company focused on the development of gene therapy products for the treatment of rare diseases, in 2011 to lead the Regulatory Science function. Prior to joining bluebird bio, Anne-Virginie was an Executive Director at Voisin Consulting, leading projects involving the design and implantation of regulatory strategies for medicinal products, with a particular focus on rare diseases, oncology, and advanced therapies.

Her experience spans from early development through commercialization, including lead roles on the registration of several orphan drugs, and regulatory expertise on both sides of the Atlantic.

Anne-Virginie holds a Master of Science in Environmental Health Sciences from the UCLA School of Public Health, as well as a Bachelor of Science in Chemical Engineering from the California Institute of Technology.

HOWARD FINGERT, M.D., Takeda Pharmaceuticals

Howard Fingert, M.D, is Senior Medical Director, Clinical Research at Takeda Pharmaceuticals, where he leads a new program in Clinical Intelligence. With both internal programs and products from external alliances, he has directed development of Oncology products designed for treatment of rare adult and pediatric indications. Dr. Fingert currently serves as Industry Representative to the Oncology Drugs Advisory Committee for the FDA. Prior to Takeda, Dr. Fingert worked in clinical development, clinical risk management, and regulatory affairs at Pfizer and other pharmaceutical & biotech companies developing products for oncology, HIV, immunology, neurology and other indications. Board-certified in Internal Medicine, Hematology and Medical Oncology, he previously served on the academic faculties at Harvard and Tufts Medical Schools, and published laboratory and clinical research in cancer pharmacology, drug safety, and phases 1-4 clinical trials.

MARYAM FOULADI, M.D., M.Sc., Cincinnati Children's

PATRICIA FURLONG, Parent Project Muscular Dystrophy

Pat Furlong is the Founding President and CEO of Parent Project Muscular Dystrophy (PPMD), the largest nonprofit organization in the United States solely focused on Duchenne muscular dystrophy (Duchenne). Their mission is to end Duchenne. They accelerate research, raise their voices in Washington, demand optimal care for all young men, and educate the global community.

Duchenne is the most common fatal, genetic childhood disorder, which affects approximately 1 out of every 3,500 boys each year worldwide. It currently has no cure.

When doctors diagnosed her two sons, Christopher and Patrick, with Duchenne in 1984, Pat didn't accept "there's no hope and little help" as an answer. Pat immersed herself in Duchenne, working to understand the pathology of the disorder, the extent of research investment and the mechanisms for optimal care. Her sons lost their battle with Duchenne in their teenage years, but she continues to fight—in their honor and for all families affected by Duchenne.

In 1994, Pat, together with other parents of young men with Duchenne, founded PPMD to change the course of Duchenne and, ultimately, to find a cure. Today, Pat continues to lead the organization and is considered one of the foremost authorities on Duchenne in the world.

JONATHAN GOLDSMITH, M.D., National Heart, Lung, and Blood Institute, NIH

Dr. Goldsmith serves as the Deputy Branch Chief, Blood Diseases Branch, Division of Blood Diseases and Resources, NHLBI, NIH.

In his career in academic medicine, he was actively involved in basic and clinical research, teaching, and patient care at four schools of medicine. His last appointment was at the University of Southern California as a Professor of Pediatrics and Medicine at Childrens Hospital Los Angeles where he directed the Childrens AIDS Center and the Region IX Hemophilia Program funded by HRSA and the CDC. Since leaving academic medicine, he has held upper management positions at three biopharmaceutical corporations, served as the Medical Director and Interim President of the Immune Deficiency Foundation, and more recently was the Deputy Director, Office of Blood Research and Review (OBRR), Center for Biologics Evaluation and Research (CBER), FDA.

Professional recognitions include designation as a Fellow of the American College of Physicians and memberships in the American Society of Hematology and other hematology societies. He also served as a board member of the National Hemophilia Foundation and as a member of the Blood Products Advisory Committee, CBER/FDA.

NANCY GOODMAN, Kids v Cancer

Nancy founded Kids v Cancer in memory of her son, Jacob, who died of medulloblastoma, a pediatric brain cancer, at age 10 in 2009.

Kids v Cancer promotes pediatric cancer research by identifying structural impediments at key junctures in the research process— access to funding, tissue and drugs – and developing strategies to resolve them. Under Nancy's leadership, Kids v Cancer has had the following accomplishments:

- The Creating Hope Act, which creates a market-based incentive to spur pediatric cancer drug development, was signed into federal law on July 9, 2012 as Section 908 of the FDA Safety and Innovation Act. Kids v Cancer authored and advocated for passage of this legislation.
- Kids v Cancer started an autopsy tissue donation program for pediatric brain cancers, which has already led to significant scientific discoveries.
- Kids v Cancer raised the profile of pediatric cancer by asking that September declared to be *National Childhood Cancer Awareness Month* for each of the past three years.
- Kids v Cancer developed an alternative application of the Best Pharmaceuticals for Children Act (BPCA) which could accelerate the development of drugs for pediatric disease and which the FDA is now implementing.

Nancy is a graduate of the University of Pennsylvania (BA), Harvard's Kennedy School of Government (MPP) The Law School at the University of Chicago (JD).

HOLCOMBE GRIER, M.D., Harvard Medical School

LEE HELMAN, M.D., National Cancer Institute, NIH

Lee J. Helman received his M.D. from the University of Maryland School of Medicine *magna cum laude* in 1980 and was elected to Alpha Omega Alpha. He completed his internship and residency in Internal Medicine at Barnes Hospital Washington University also serving as Chief Resident. Dr. Helman began his fellowship training at the National Cancer Institute (NCI) in 1983, where he has remained. He did his post-doctoral training in the Molecular Genetics Section, Pediatric Branch, NCI, and became Head of the Molecular Oncology Section, Pediatric Oncology Branch, NCI, in 1993. He served as Chief of the Pediatric Oncology Branch from 1997-2007, and in 2007 became Scientific Director for Clinical Research in the Center for Cancer Research, National Cancer Institute, a position he currently holds. Dr. Helman is a Professor of Pediatrics and Oncology at the Johns Hopkins University. He was elected to the American Society for Clinical Investigation and the American Association of Physicians and is a founding member and past president of the Connective Tissue Oncology Society. He serves as Chairman of the Board of Directors of and is a Clinical Advisor to The Children's Inn at NIH and is a past member of the Board of Governors of the Clinical Center at NIH. Dr. Helman is a

past member of the Board of Directors of the American Society of Clinical Oncology (ASCO) and a past chair for its Bylaws Committee. He received the 2011 ASCO Pediatric Oncology Award and the ASCO Statesman Award. He has served on the Science Education, Publications, and Clinical & Translational Research committees of the American Association of Cancer Research (AACR) and is chair of its Pediatric Oncology Task Force and has been on the Scientific Program Committee for several of its annual meetings. He serves on the Scientific Advisory Committee of Stand Up To Cancer, a scientific partner to the AACR. He is on the Scientific Advisory Committee of the Children's Oncology Group. Dr. Helman served as an associate editor for the journal *Cancer Research* and *Clinical Cancer Research* and currently is on the editorial board of the *Journal of Clinical Oncology*. Dr. Helman's laboratory currently focuses on three major themes related to the biology and treatment of pediatric sarcomas, specifically Ewing's sarcoma, rhabdomyosarcoma, and osteosarcoma: (1) determine the pathophysiologic consequences of IGF signaling; (2) identify the molecular/biochemical determinants of the biology of these sarcomas; and (3) apply preclinical laboratory findings to develop novel clinical studies for these sarcomas.

KATHERINE HIGH, M.D., Univ. PA School of Medicine; Howard Hughes Medical Institute; and Children's Hospital of Philadelphia

Katherine High was born in High Point, North Carolina on July 27, 1951. She graduated from Harvard with a degree in Chemistry in 1972. She then carried out her M.D. studies at the University of North Carolina (UNC) School of Medicine, graduating in 1978. After completing training in internal medicine, she trained as a Fellow in the Hematology Section at Yale University School of Medicine from 1981-84. After a year on staff at Yale, she returned to UNC as a faculty member in the Department of Medicine and the Curriculum in Genetics from 1985-1992. In 1992, she moved to Philadelphia where she is presently William H. Bennett Professor of Pediatrics at the University of Pennsylvania School of Medicine, Investigator, Howard Hughes Medical Institute, and Director, Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia.

Dr. High's research interests focused initially on the molecular basis of blood coagulation. She characterized a number of gene mutations that lead to hemophilia B, and to deficiencies of Factor X and Factor VII, and used recombinant expression systems to produce large amounts of clotting factor proteins for detailed biochemical studies. She was the first to clone a canine gene, when she isolated the gene encoding canine Factor IX; this paved the way for her subsequent elucidation of the mutation that leads to canine hemophilia B. She has been a pioneer in the development of novel genetic therapies to treat inherited disorders, and in particular has been at the forefront of safe and effective clinical translation of genetic therapies for hemophilia. She led the first human trial of introduction of AAV vectors into skeletal muscle, the first trial of administration of AAV vectors to liver, and the first trial in the U.S. of introduction of AAV into the subretinal space. These clinical trials have led to correction of disease in hemophilia B, and in Leber's congenital amaurosis, a hereditary cause of blindness. Her rigorous analysis of problems uncovered in the course of clinical studies, including risk of germline transmission of vector in recipients of donated DNA, and of immune responses to viral vectors following

delivery to specific tissues, has laid the foundation for continued expansion of the applications of gene therapy.

Dr. High is the past recipient of the National Hemophilia Foundation Researcher of the Year Award (2000), and was elected as a AAAS Fellow for "Distinguished contributions in the field of human gene therapy" in 2000. She received the Stanley N. Cohen Biomedical Research Award from the University of Pennsylvania in 2001, the 2003 Distinguished Alumna Award from the University of North Carolina at Chapel Hill, and a Distinguished Achievement Award from the American Heart Association, ATVB Council in 2004. She received the Foundation for Fighting Blindness Board of Directors Award in 2007, the Investigator Recognition Award from the International Society of Thrombosis and Hemostasis 14th Biennial Awards for Contributions to Hemostasis in 2009, and is a co-recipient of the 2009 Audrey E. Evans Award of Excellence from the Philadelphia Ronald McDonald House. She is also the recipient of the 2010 Outstanding Achievement Award from the American Society of Gene and Cell Therapy.

Dr. High has published over 150 scientific articles, including the first reports of AAV-mediated gene transfer into muscle and liver in patients with hemophilia B, and into the retina in patients with an inherited retinal disorder that causes blindness in childhood. She is an editor of a hematology textbook, Clinical Hematology.

Dr. High is an active member of the ISTH. She is a member of the American Society of Hematology, and of the American Society of Gene and Cell Therapy, of which she is a Past President (2004-05). She has been elected to a number of honorary societies including the American Society for Clinical Investigation, the Association of American Physicians, the Institute of Medicine of the National Academies, and the American Academy of Arts and Sciences. Dr. High served on the National Heart, Lung, and Blood Institute (NHLBI) Advisory Council, the NHLBI Gene Therapy Resource Program (GTRP) Steering Committee, and the FDA Cellular, Tissue and Gene Therapies Advisory Committee (CTGAC). She served on the Scientific Advisory Board of the Italian Telethon Foundation. She is an associate editor of Molecular Therapy, and serves on the editorial boards of Human Gene Therapy and Blood. In 2005, Dr. High became the founding director of the Center for Cellular and Molecular Therapeutics of The Children's Hospital of Philadelphia, which has as its mission the development of novel cell and gene therapies for inherited disorders that affect children.

Dr. High serves on the Scientific Advisory Boards of bluebird bio and of Alnylam Pharmaceuticals. She is a Co-founder of Spark Therapeutics.

STEVEN HIRSCHFELD, M.D., Ph.D., National Institute of Child Health and Human Development, NIH

Captain Steven Hirschfeld of the Commissioned Corps of the United States Public Health Service (USPHS) earned his MD from Columbia University College of Physicians and Surgeons and his PhD in Cell Biology from New York University. He did his residency and Chief Residency at the University of California, San Francisco, research fellowship training at the

Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), and clinical fellowship at the National Cancer Institute (NCI), National Institutes of Health, Bethesda, MD.

Dr. Hirschfeld received board certification in general pediatrics and pediatric hematology-oncology. He worked at the National Cancer Institute as a clinical investigator and then at the Food and Drug Administration (FDA) in the Center for Drug Evaluation and Research and the Center for Biologics Evaluation and Research. He contributed to the development and implementation of the federal pediatric initiatives beginning in 1996, the International Conference for Harmonization E11 Pediatric Guidance, and the adaptation of regulations for the protection of children in FDA regulated research. In 2006 he returned to NICHD as Associate Director for Clinical Research and also serves as NIH Co-Coordinator of the Child Health Oversight Committee for the National Center for Research Resources Clinical and Translational Science Awards Consortium. In addition to regulatory, policy, and operations activities, Dr. Hirschfeld has been active in the development and implementation of data standards for acquisition, transmission, and analysis as well as exploring new methods of data analysis. He is the recipient of numerous awards, including twice the Health and Human Services Secretary's Award for Distinguished Service and awards from the Commissioner of the Food and Drug Administration, the Director of the National Institutes of Health, and the USPHS.

Dr. Hirschfeld has deployed regularly on humanitarian and disaster relief missions with the USPHS since 1991, including two deployments to Louisiana in 2005 for the Gulf Coast Hurricanes and in 2008 to Texas for Hurricane Ike. He serves as the Chief Medical Officer and Operations Chief for the USPHS Rapid Deployment Force Team -1 based in the Washington, DC metro area.

ROBERT IANNONE, M.D., M.S.C.E., Merck and Co., Inc.

I received a BS degree in Biology from The Catholic University of America in 1989 with Summa Cum Laude and Phi Beta Kappa honors and subsequently graduated from Yale University School of Medicine in 1994, where I was elected to Alpha Omega Alpha. My MD thesis work was on human T cell leukemia virus under the mentorship of Dr. Clarence J. Gibbs (NINDS, NIH). After internship and residency in Pediatrics at Johns Hopkins Hospital, where I also spent an additional year as Chief Resident, I completed a fellowship in Pediatric Hematology and Oncology in 2001 and currently maintain board certification in Pediatrics and Pediatric Hematology-Oncology. My research during fellowship was under Dr. Ephraim Fuchs in the Division of Cancer Immunology and Hematopoiesis at Kimmel Cancer Center at Johns Hopkins. This research on immune tolerance in bone marrow transplantation led to a novel clinical bone marrow transplant trial in patients with sickle cell disease.

I was appointed to the standing faculty of the University of Pennsylvania Medical School in 2001 at the rank of Assistant Professor. While on faculty, I was an attending physician in Pediatric Oncology with an emphasis in pediatric bone marrow transplantation at Children's Hospital of Philadelphia (CHOP). My NIH-funded research interest involved translational

clinical trials in bone marrow transplantation related to sickle cell disease, inborn errors of metabolism and hematologic malignancies. During that time, I enrolled in the Master's Program in Clinical Epidemiology (MSCE) at the Center for Clinical Epidemiology and Biostatistics at the University of Pennsylvania Medical School, and ultimately was awarded this degree. I am currently an Adjunct Assistant Professor of Pediatrics and regularly attend Pediatric Oncology Clinic.

I was recruited to Merck Clinical Pharmacology in December 2004 as an Associate Director and was promoted to Director in November, 2006. While in Clinical Pharmacology, I was directly involved in a number of drug development programs across several of therapeutic areas, including neuroscience, oncology, hematology, biologics, respiratory and cardiovascular. I was the program chair for two of these teams and, during that time, completed more than 20 phase I clinical trials, including 2 phase Ib proof-of-concept and 3 PET imaging studies. I also contributed to phase II studies and supported numerous regulatory interactions.

In April of 2008 I was promoted to Senior Director and Site Head for Experimental Medicine in Upper Gwynedd, where I oversaw the research groups for Experimental Medicine Oncology, Neuroscience and RNA Therapeutics. This involved direct oversight for clinical translational research protocol development and clinical trial execution. During 2010, I lead the integration effort in Early Stage Development after the merger with Schering-Plough.

As of March 1, 2011 I was appointed Section Head in Clinical Oncology. I am currently the clinical lead for the MK-3475 New Indications and Immuno-modulatory Regulator (IMR) teams. I continue to serve as co-Chair for the Pediatric Development Committee.

ILAN IRONY, M.D., Center for Biologics Evaluation and Research, FDA

Dr. Irony is an internist and endocrinologist, who joined the FDA (CBER) in September of 2000 as a clinical reviewer after 6 years in group medical practice in the Washington DC area. He transferred to the Division of Metabolism and Endocrinology Products in CDER in 2005, and became a clinical team leader in 2009. Since December 2011, he has been back to CBER as the chief of the General Medicine Branch, DCEPT / OCTGT. Dr. Irony wrote FDA draft and final guidances, and actively participates in scientific and regulatory working groups within and outside FDA. He has extensive experience in reviews of products for rare diseases, and currently leads the General Medicine Branch where a substantial effort is devoted to the clinical review of cellular and gene therapy products intended to treat rare diseases.

JESSICA LEE, M.D., M.M.Sc., Center for Drug Evaluation and Research, FDA

Jessica Lee is a medical team leader in the Division of Gastroenterology and Inborn Errors Products (DGIEP) in the Center for Drug Evaluation and Research (CDER) at FDA. She received her undergraduate degree from the Massachusetts Institute of Technology and her medical degree from the State University of New York Upstate Medical University. She

completed her residency training in pediatrics at Children's Hospital at Montefiore, Albert Einstein College of Medicine, followed by fellowship training in pediatric gastroenterology at Boston Children's Hospital. She also completed the Scholars in Clinical Science Program, a two-year postgraduate training program in clinical investigation at Harvard Medical School. She was a faculty at Boston Children's Hospital and Harvard Medical School prior to joining the FDA as a Medical Officer in 2011. She currently leads a medical review team within DGIEP that primarily focuses on review of investigational and new drug applications for rare, inherited metabolic diseases.

CYNTHIA LE MONS, National Urea Cycle Disorders Foundation

Cynthia Le Mons is Executive Director and CEO of the National Urea Cycle Disorders Foundation (NUCDF). Le Mons began her commitment to NUCDF over 18 years ago when a family member was diagnosed with urea cycle disorder (UCD), serving as a volunteer, board member, and later as president. Under her leadership, NUCDF has grown to be an internationally-respected patient advocacy organization leading research efforts to improve the understanding of UCD and driving the development of new interventions and treatments. She has held leadership roles in many national collaborative efforts on behalf of children with inborn errors of metabolism, including state-mandated newborn screening and acceleration of translational research to lessen the burden of disease, improve survival and neurocognitive outcome. She has led NUCDF in a model partnership with researchers to develop the National Institutes of Health Rare Diseases Clinical Research Network (RDCRN) Urea Cycle Disorders Consortium to catalyze critical research and focus translational research studies for urea cycle disorders. She is actively involved in research protocol development and leading strategic direction for the consortium. Le Mons also serves as co-chair of the NIH RDCRN Coalition of Patient Advocacy Groups and is a member of the RDCRN steering committee. Her commitment to fostering model collaborations with industry has led to successful FDA orphan drug approvals for urea cycle disorders and a robust pipeline for future clinical trials.

PHILIP MARELLA, Niemann-Pick type C disease

Phil Marella is a Trustee of Dana's Angels Research Trust, or DART, an all-volunteer 501(c)(3) public charity founded in 2002 by Mr. Marella and his wife, Andrea, when their daughter, Dana, then age 8, was diagnosed with the rare, degenerative disease, Niemann-Pick type C (NPC). Tragically, Dana passed away this past summer just 11 days short of her 20th birthday. NPC is a disorder that prohibits the proper metabolism of cholesterol and other lipids. The Marella's youngest child, Andrew, age 14, is also afflicted with NPC. DART works to fund NPC research with the National Niemann-Pick Disease Foundation and the Ara Parseghian Medical Research Foundation, and is particularly proud of being a founding member in a unique, multidimensional collaborative drug development program called SOAR-NPC for Support Of Accelerated Research for NPC.

Professionally, Mr. Marella is President of Green Light Worldwide Media, Inc., a film and television development, production and management company founded in 2000. Mr. Marella has over 30 years of experience with financial and legal management in the film and television business. Before joining Green Light, Mr. Marella was a senior business affairs and legal executive for different media companies including Microcast, Inc. and Worldvision Enterprises, Inc., the global marketing and distribution subsidiary of Spelling Entertainment Inc. Mr. Marella began his career in various positions in finance and accounting at Capital Cities/ABC, Inc. Mr. Marella has a law degree from Fordham University School of Law and a Bachelor of Science degree with Special Attainments in Commerce from Washington and Lee University.

D. ELIZABETH MCNEIL, M.D., M.Sc., National Institute of Neurological Disorders and Stroke, NIH

Dr. Elizabeth McNeil joined NINDS in January 2011 after working for 8 years at the US Food and Drug Administration (FDA) as a medical reviewer and as a clinical team leader. She did her undergraduate work at the University of Chicago. She subsequently graduated from Columbia University, College of Physicians and Surgeons. She trained in pediatrics at Texas Children's Hospital before training in neurology, pediatric neurology and neuro-oncology at the Children's Hospital of Philadelphia. She is board certified in Pediatric Neurology. After completion of fellowship training, she was in private practice before going to a second fellowship in genetic epidemiology at the National Cancer Institute of the National Institutes of Health. Dr. McNeil has a Master of Science in Epidemiology from the University of London, UK.

While at the FDA, she worked extensively on a full range of products, including new molecular entities, new formulations of approved drug products, drug-drug combinations, and drug-device combinations. She worked to develop new scientific, clinical and regulatory standards for the development of certain drug classes and developed initiatives which resulted in standardization and clarification of terminology used in defining claims and endpoints, as well as in increased standardization, precision, and sensitivity in reporting safety and efficacy data.

At NIH/NINDS, she is scientific program director for the new Network for Excellence in Neuroscience Clinical Trials (Neuro-NEXT, www.NeuroNEXT.org).

She is a member of the IRDiRC Therapies Scientific Committee. She also serves as government liaison to multiple advocacy groups' drug development boards.

JANA MONACO, Organic Acidemia Association & Children's National Medical Center

Ms. Monaco is the mother of two children with Isovaleric Acidemia including one child with severe neurological impairment and complex health issues due to late diagnosis and one affected child that was tested in the newborn period and provided treatment to prevent complications. Through her experiences, she has been an advocate in the rare disease community for issues such

as newborn screening, medical foods insurance coverage, the medical home and neurodevelopmental disabilities awareness.

Jana has served as a member of the Secretary of Health and Human Services Advisory Committee for Heritable Disorders in Newborns and Children and is currently a member of its Follow Up and Treatment Subcommittee. As a member of the Virginia Genetics Advisory Committee, Jana has supported legislation for expanded newborn screening which was implemented in the State of Virginia on 1 March 2006. She is the Advocacy Liaison for the Organic Acidemia Association providing support to families with children affected with inborn errors of metabolism.

She served as the Family Faculty Member of the Leadership Education and Training in Neurodevelopmental Disabilities Program (LEND) at Children's National Medical Center and is a member of the hospital Patient/Family Advisory Council currently serving as the chair of the council, whose mission is to ensure that patient and family centered care is incorporated in every patient experience.

Jana's advocacy has included presentations on the topic of Newborn Screening to the Board of Directors of the March of Dimes, the Northern Virginia Pediatric Society, Inova Fairfax Hospital Residents, the National Organization for Rare Diseases (NORD) and Genetic Alliance Conferences and the renaming ceremony for the Eunice Kennedy Shriver National Institute for Child Health and Human Development. She has participated in panel discussions to include the March of Dimes Congressional Briefing on Medicaid and Children and the 50th Anniversary of Newborn Screening on Capitol Hill. She is published in Exceptional Parent Magazine, "My Poster Family" and given media interviews on newborn screening to The Wall Street Journal, Newsweek Magazine, The Potomac News, CBS, NBC and ABC

ANDREW MULBERG, M.D., F.A.A.P., C.P.I., Center for Drug Evaluation and Research, FDA

Andrew is currently the Division Deputy Director of Gastroenterology and Inborn Errors Products, Center for Drug Evaluation and Research (CDER), U.S. Food and Drug Administration (FDA) since 2010. Before joining FDA, Andrew has served as Portfolio Leader in Established Products responsible for providing worldwide leadership in support of GI and diverse Internal Medicine products within the Established Products Therapeutic Area of Johnson and Johnson from 2000-2010. He has served as Attending Physician in Gastroenterology and Hepatology at Children's Hospital of Philadelphia from 1993-2010. Andrew is a graduate of Columbia College of Columbia University and of the Mount Sinai School of Medicine. He completed his residency in Pediatrics at the Children's Hospital of Philadelphia followed by a Pediatric Gastroenterology Clinical Fellowship and a Post-Doctoral Fellowship in Cellular and Molecular Physiology at New England Medical Center. Andrew is Adjunct Professor of Pediatrics at the University of Maryland School of Medicine ,Adjunct Associate Professor of Pediatrics in the University of Pennsylvania School of Medicine and Associate Professor of Pharmacy at the University of the Sciences in Philadelphia. He has served as Principal Editor of

Pediatric Drug Development: Concepts and Applications published April 2009 with Wiley-Blackwell and now in its 2nd edition released August 19, 2013. He is a member of multiple professional medical societies including Alpha Omega Alpha Honor Medical Society, American Gastroenterological Association and the North American Society for Pediatric Gastroenterology and Nutrition.

DIANNE MURPHY, M.D., F.A.A.P., Office of the Commissioner, FDA

Dr. Dianne Murphy has been the Director of the Office of Pediatric Therapeutics in the FDA's Office of the Commissioner since 2004. Previously, in the center for Drug Evaluation, Dr. Murphy was Director of the Office of Counter-terrorism and Pediatric Drug Development (2001-2004), the Associate Director for Pediatrics (1998-2001), and Director of the Office of Drug Evaluation with oversight for all of the Divisions involved with antimicrobial therapeutics (1998-2001).

Dr. Murphy received her medical degree from the Medical College of Virginia. After completing a pediatric residency at the University of Virginia, she spent three years at the National Naval Medical Center as a Navy pediatrician before completing a fellowship in pediatric infectious diseases at the University of Colorado. Dr. Murphy was an Assistant Professor for Pediatrics at the University of Texas Health Science Center at San Antonio, an Associate Professor of Pediatrics and medical consultant to the Diagnostic Virology Laboratory at the University of Tennessee Medical Center at Knoxville and Professor of Pediatrics and Chief of General Pediatrics at the University of Florida Health Science Center at Jacksonville. Dr. Murphy has numerous articles in refereed publications on pediatric infectious diseases, pediatric drug development, residency teaching, and laboratory diagnosis and is the editor of a book on Pediatric Drug Development and Office Laboratory Procedures.

The Office of Pediatric Therapeutics has legislatively mandated responsibilities for all of FDA's pediatric activities with a particular focus on safety and ethics. Ethically sound pediatric trials must be scientifically sound and because pediatric trials are often global, the office also coordinates international pediatric activities and exchanges of information related to pediatric trial issues. She received the American Academy of Pediatrics: Excellence in Public Service Award in 2013.

ROBERT "SKIP" NELSON, M.D., Ph.D., Office of the Commissioner, FDA

Robert "Skip" Nelson, M.D., Ph.D. is currently the Deputy Director and Senior Pediatric Ethicist in the Office of Pediatric Therapeutics, Office of the Commissioner at the U.S. Food and Drug Administration. Immediately prior to joining FDA full-time in 2009, he was Professor of Anesthesiology, Critical Care and Pediatrics at The Children's Hospital of Philadelphia and University of Pennsylvania School of Medicine. After receiving his M.D. degree from Yale University, Dr. Nelson trained in pediatrics (Massachusetts General Hospital), neonatology and pediatric critical care (University of California, San Francisco). He has a Master of Divinity

degree from Yale Divinity School and a Ph.D. in The Study of Religion from Harvard University.

Dr. Nelson was a member (2004-2006) and former Chair (2005-2006) of the FDA Pediatric Advisory Committee and the Pediatric Ethics Subcommittee. He was a member of the Subcommittee on Research Involving Children of the Secretary's Advisory Committee on Human Research Protections (2003-2006), and the Human Studies Review Board of the Environmental Protection Agency (2006). Dr. Nelson was a member of the Committee on Clinical Research Involving Children of the Institute of Medicine (2002-2004), and a member and former Chair of the Committee on Bioethics of the American Academy of Pediatrics (1994-2001). Dr. Nelson is the Editor-in-Chief of the AJOB – Empirical Bioethics, which publishes empirical research in bioethics. Dr. Nelson's academic research explored various aspects of child assent and parental permission, including risk perception and voluntary choice, and was funded by the Greenwall Foundation, the National Institutes of Health, and the National Science Foundation.

DANIEL ORY, M.D., Washington University School of Medicine

Dr. Ory graduated from Harvard College and Harvard Medical School and received his scientific training at the Whitehead Institute for Biomedical Research at MIT. He joined the faculty at Washington University, St. Louis in 1995 and is currently the Alan A. and Edith L. Distinguished Professor of Medicine. Dr. Ory's research program is centered on cholesterol biology and Niemann-Pick disease type C (NPC) disease, a pediatric, neurodegenerative cholesterol storage disorder. He has established an internationally recognized research program that has made important contributions to our understanding of NPC1 protein function and its role in regulating cellular cholesterol. The Ory laboratory was also the first to recognize that the major human NPC1 disease allele, I1061T, produces a misfolded, yet functional protein, which has opened up investigation of potential proteostatic therapies, such as histone deacetylase inhibitors, for NPC1 disease. In 2008, in partnership with NPC families he founded SOAR-NPC, an international collaborative group whose goal is to identify NPC disease biomarkers and develop new therapies for NPC disease. His work has led to development of new diagnostic biomarkers for NPC and clinical assays that are being implemented worldwide. Since 2010, Dr. Ory has worked closely the NIH Therapies for Rare and Neglected Diseases (TRND) program to develop a Phase 1 trial for intrathecal delivery of cyclodextrin that was initiated at the NIH Clinical Center in January 2013, and is leading the design of the Phase 2/3 trial. Dr. Ory has served on the Scientific Advisory Board of the National Niemann-Pick Disease Foundation since 2002, and as Chair since 2007. He holds numerous university appointments, including Co-Director of the Diabetic Cardiovascular Disease Center, Director of Admissions for the Division of Biology and Biomedical Sciences, Associate Director of the Medical Scientist Training Program, and Director of the Metabolomics Facility.

MARK PAPIER, Niemann-Pick Disease Type C

Mark Papier, his wife Darrile, and their 11 year old son, Dillon, who was diagnosed with Niemann-Pick Disease Type C, at three years of age live in Frederick, Maryland. Mark has been employed as a high school psychology/sociology teacher, in the Frederick Maryland school system, spanning the last 32 years.

Mark has been a JV Baseball Coach for the past 23 years. He has brought this same love of baseball and everything related to the Baltimore Orioles and Washington Nationals to his, son, Dillon, their biggest fan.

Since Dillon's diagnosis, the Papier family has been dedicated advocates on behalf of their captivating and energetic son. Realizing that time was NOT on their side and understanding the desperate need for new therapy options associated with NPC, the family stepped outside of the comfort zone and became passionate advocates on behalf of their son.

On the patient advocacy front and in support of all NPD Type C patients, Mark, Darrile, their extended family, as well as the wider community of Frederick, have worked tirelessly to raise the funds necessary to drive NPC research forward to ensure new therapies and treatment options for Dillon and other NPD patients. They have also been involved with the NPD Foundation, the Board of Directors, and served as host family of the Annual NNPDF Family Support and Medical Conference and to members of the International Niemann-Pick Disease Alliance (INPDA) in August 2013 in Baltimore, MD.

Currently, there is no FDA approved therapy or treatment for NPD Type C. After exhaustive research, in-depth discussions and soul searching, Mark and Darrile made the courageous decision to enroll Dillon in the current FDA approved NIH/TRND Cyclodextrin pediatric clinical trial taking place at the National Institutes of Health.

GAIL PEARSON, M.D., Sc.D., F.A.C.C., F.A.H.A., F.A.A.P., National Heart, Lung, and Blood Institute, NIH

Gail Pearson joined NHLBI in 1997. A board-certified pediatric cardiologist, she received her MD from Johns Hopkins in 1991 as well as an ScD in health economics and health policy from the Johns Hopkins School of Hygiene and Public Health in 1990. She completed her pediatric residency and pediatric cardiology fellowship at Children's National Medical Center, with an emphasis on echocardiography. Since coming to NHLBI, Gail has designed and launched the Pediatric Heart Network (2001), and more recently, the Bench to Bassinet program, a comprehensive translational research program in pediatric cardiovascular diseases. Gail was part of the team that developed the award-winning Children and Clinical Studies web site and campaign to inform parents about clinical research. Gail is also the Director of the NHLBI Office of Clinical Research, which develops and implements policy for the oversight of NHLBI-funded clinical research. Gail continues to see patients and perform non-invasive imaging at Children's National Medical Center.

HOLLY PEAY, M.S., C.G.C., Parent Project Muscular Dystrophy

Holly Peay is the Vice President of Outreach and Education for Parent Project Muscular Dystrophy (PPMD), the largest Duchenne-focused advocacy group. In addition, she directs the DuchennneConnect registry. Her effort is shared between education programs (to families, healthcare providers and the public) and research conducted through PPMD. She also participates in social/behavioral research as a Guest Researcher/Personal Services Contractor at the NHGRI. Currently, Mrs. Peay is the PI of an NINDS-funded study on therapeutic misconception and clinical trial expectations in the DBMD community, and her ongoing research includes a study of predictors of adaptation in mothers of individuals with Duchenne and Becker muscular dystrophy, and a program on Duchenne treatment priorities and risk tolerance.

Mrs. Peay has a Bachelors in Biomedical Ethics from the University of Virginia and a Masters in Genetic Counseling from the University of South Carolina School of Medicine, and a PhD is anticipated Summer 2014 from Leiden University Medical Centre.

BETSY PETERSON, The Children's Heart Foundation

Betsy Peterson is the founder of The Children's Heart Foundation, the national non-profit organization started in 1996 after the death of the Peterson's eight-year-old son Sam from congenital heart defects. CHF raises money to fund the most promising research to advance the diagnosis, treatment, and prevention of congenital heart disease. To date CHF has granted over $6 million to 56 research studies, laying the groundwork for many to receive funding later on from the National Institutes of Health. In 2012 she partnered with filmmaker Ted Kay to create the one-hour documentary "The Heart of the Matter" on congenital heart disease. So far, more than 80% of PBS stations nationwide have broadcast this program which will be available on-line in early 2014 at www.congenitalheartdocumentary.com. She has participated in past NHLBI Public Information Gatherings to share information with other non-profits on partnerships, clinical research support, and awareness. She lives in Chicago with her husband, playwright Steven Peterson. They are the parents of a daughter, Hallie. Betsy holds a B.A. in communications from St. Cloud State University, and has held past positions in fund-raising and public relations for The Illinois Institute of Technology, The University of Chicago Children's Hospital and the American Cancer Society.

FORBES PORTER, M.D., Ph.D., Eunice Kennedy Shriver National Institute of Child Health and Human Development, NIH

Dr. Porter is a Senior Investigator and Program Head in the intramural research program of the Developmental Endocrinology and Genetics Program (PDEGEN) of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD). Dr. Porter also serves as the Clinical Director for NICHD. Dr. Porter received both his MD and PhD degrees from

Washington University in St. Louis, and trained in both Pediatrics and Clinical Genetics at St. Louis Children's Hospital. He then moved to the NIH as a postdoctoral fellow in 1993, and started his own research group in 1996. His research program studies basic, translational, and clinical aspects of genetic disorders with impaired cholesterol homeostasis. These include Smith-Lemli-Opitz syndrome and Niemann-Pick Disease, type C. The goal of this research program is to understand pathophysiogical processes underlying these disorders, develop therapeutic interventions, and implement therapeutic trials. Dr. Porter works closely with patient advocate groups. He is on the scientific/medical advisory boards of both the RSH/Smith-Lemli-Opitz Foundation and the National Niemann-Pick Disease Foundation.

GREGORY REAMAN, M.D., Center for Drug Evaluation and Research, FDA

Gregory H. Reaman, M.D. joined the Center for Drug Evaluation and Research, Office of New Drugs, U.S. Food and Drug Administration as the Associate Director of the Office of Oncology Drug Products in 2011. He is the Founding and Immediate Past Chair of the Children's Oncology Group (COG) having served in this capacity from 2000 through 2010. The COG is comprised of over 200 member institutions, dedicated to clinical, translational, and epidemiology research in childhood cancer.
Dr. Reaman is a Professor of Pediatrics at The George Washington University School of Medicine and Health Sciences and a member of the Division of Hematology-Oncology at the Children's National Medical Center in Washington, D.C., which he directed for more than 17 years and Executive Director Emeritus of the Center for Cancer and Blood Disorders.

Dr. Reaman serves or has served on the Editorial Boards of Leukemia, Journal of Clinical Oncology, Journal of Pediatric Hematology/Oncology, Pediatric Blood and Cancer, The Oncologist, Cancer, and Physicians Data Query (PDQ), National Cancer Institute as well as www.PLWC.org (People Living with Cancer, now Cancer.net). He has served as an Associate Editor of Cancer and Leukemia and Lymphoma. He held the position of Executive Director for Scientific and Medical Affairs for the National Childhood Cancer Foundation (NCCF) and was a member of its Board of Trustees. Previously, he served on the Board of Directors of the American Cancer Society and chaired its Task Force on Children and Cancer. Dr. Reaman served on the Board of Directors of the American Society of Clinical Oncology (ASCO)and has served on the ASCO Patient Education Committee, the Education and Program Committees, the Cancer Research Committee, the Grant Selection Committee, Cancer Survivorship Committee, and served as Chair of the ASCO Membership and Audit Committees. Also, he was a member of the Food and Drug Administration's Oncologic Drugs Advisory Committee and has chaired its Pediatric Subcommittee. He was a member of the NIH Roadmap Working Group. He serves on the Scientific Steering Committee of the United Kingdom Children's Cancer and Leukemia Group, the External Advisory Board of the Cancer Treatment and Research Center at the University of Texas Health Science Center at San Antonio and is a Senior Advisor to the Middle East Childhood Cancer Alliance (MECCA).

Additionally, he is a member of the Alliance for Childhood Cancer, a member of the Data Safety Monitoring Board of the National Cancer Institute's Clinical Oncology Program, and a member of the NCI's Translational Research Working Group.

His research interests are in the biology and treatment of childhood acute leukemia and new drug development for pediatric cancers.

He is the author of more than 300 peer - reviewed manuscripts and 16 book chapters.

LORI SAMES, Hannah's Hope Fund

For the last five and a half years, Lori has been managing a virtual biotech, leading a collaborative team of scientists to focus on developing a treatment for Giant Axonal Neuropathy (GAN). Lori and her husband created Hannah's Hope Fund, a 501c3 public charity, in 2008 after their 4 year old daughter received this devastating diagnosis.

GAN is the most severe form of inherited neuropathy and in most cases is fatal in the early 20's. Hanna't Hope Fund has raised more than $6mn, grass-roots, for the development of GAN gene delivery to the CNS, as well as for the discovery of the molecular target(s) in GAN.

Just 3 years and 3 months after the gene delivery project began, they held their Pre Investigational New Drug Meeting with the FDA CBER. They obtained RAC approval on June 12th and plan to submit to FDA CBER and the IRB next month.

Children and young adults with GAN will likely be the first disease community in the world to receive a therapeutic gene to the spinal cord.

Prior to co-founding Hannah's Hope Fund, Lori was a stay-at-home mom, having put her career aside as a Senior Project Leader, overseeing software installations at integrated healthcare delivery networks nationally.

MALCOLM SMITH, M.D., Ph.D., National Cancer Institute

Dr. Smith is Associate Branch Chief, Pediatrics in the Cancer Therapy Evaluation Program (CTEP), NCI. Dr. Smith has been a member of CTEP since 1990 and during his years at CTEP has focused on developing NCI's preclinical and clinical research programs for children with cancer.

Dr. Smith serves as the Program Director and primary NCI liaison to childhood cancer researchers in the Children's Oncology Group (COG), focusing primarily on hematologic malignancies and brain cancers. He also serves as the Program Director for the COG Phase 1/Pilot Consortium and for the Pediatric Brain Tumor Consortium, and he is the NCI Project Officer for the Pediatric Preclinical Testing Program. Dr. Smith served as NCI co-leader of the

Childhood Cancer TARGET Initiative, a program using modern genomics technologies to identify childhood cancer genomic alterations with potential clinical impact. He serves as a member of the NCI PDQ Pediatric Editorial Board, the Editorial Board of *Pediatric Blood and Cancer*, and the Discovery and Development Committees of the NCI Experimental Therapeutics (NExT) Program, which review agents proposed for NCI preclinical and clinical development.

Dr. Smith has graduate degrees from Harvard University (M.A.) and Washington University (Ph.D.). He obtained his M.D. degree from the Medical College of Georgia and completed his Pediatric Residency training at Geisinger Medical Center and his Pediatric Hematology/Oncology fellowship training at the Pediatric Branch of the National Cancer Institute. Dr. Smith is Board certified in Pediatrics and in Pediatric Hematology-Oncology.

Dr. Smith's NCI achievements have been recognized by five Public Health Service Commendation Medals and by the NCI Director's Award and the NIH Director's Award. He is the author/coauthor of more than 100 original publications in the field of childhood cancer and clinical trials. He has also authored 25 book and monograph chapters, and he served as an editor for the *Cancer Incidence and Survival among Children and Adolescents: United States SEER Program 1975-1995* monograph.

BRENDA WEIGEL, M.D., M.Sc., University of Minnesota Amplatz Children's Hospital

Dr. Brenda Weigel is a Pediatric Hematologist/Oncologist at the University of Minnesota Amplatz Children's Hospital. Dr. Weigel received her medical degree from McMaster University in Hamilton, Ontario. She came to the University of Minnesota in 1996 to do her fellowship. During her fellowship, Dr. Weigel worked in the lab of Dr. Bruce Blazar and developed a mouse model in which to study a rare cancer called rhabdomyosarcoma.

Dr. Weigel is currently the Director of the Division of Pediatric Hematology/Oncology. She is an associate professor cross-appointed at the University of Minnesota's Cancer Center and the Department of Pediatrics, and the recipient of the Lehman/Children's Cancer Research Fund Endowed Chair in Pediatric Cancer. She is also the Co-Director of the Sarcoma Program for the Masonic Cancer Center, and Medical Director of the Clinical Trials Office of the Masonic Cancer Center.

Dr. Weigel's translational research interests are centered in her extensive involvement with the Children's Oncology Group (COG). She serves as Chair of the Developmental Therapeutics Committee of the COG leading the development of new therapies for children with cancer nationally. Her major interests are sarcoma (tumors of bone and muscle) and advancing new therapies, particularly therapies that harness the immune system's role in fighting cancer.

SUSAN WEINER, Ph.D., Children's Cause for Cancer Advocacy

Susan L. Weiner, Ph.D. is president and founder of The Children's Cause for Cancer Advocacy, a leading national education and advocacy group, dedicated to accelerating the discovery of effective treatments for childhood cancer and ensuring ready access to services for patients, families and survivors. Dr. Weiner received a Ph.D. from Columbia University in cognitive developmental psychology and completed an NIH post-doctoral fellowship. Following the diagnosis of brain tumor in her infant son, and throughout his 13 years of life, Dr. Weiner started programs for children with disabilities and their families, including founding an independent school for children with learning disabilities in New York City. She was the first executive director and continues as a board member of the Children's Brain Tumor Foundation.

Dr. Weiner has served as a childhood cancer patient and family advocate at the Institute of Medicine, the Secretary's Advisory Committee of Human Research Protections, the Association for the Accreditation of Human Research Protection Programs, the Food and Drug Administration, and National Cancer Institute planning and advisory committees. She has testified before Congress, the President's Cancer Panel and has represented patients and families in the Children's Oncology Group, the Pediatric Brain Tumor Consortium, the Cancer Leadership Council, the Alliance for Childhood Cancer, the American Society of Clinical Oncology and the American Association for Cancer Research.

DAVID WILLIAMS, M.D., Boston Children's Hospital & Harvard Medical School

Dr. Williams is the Chief of Hematology/Oncology and Director of Translational Research at Boston Children's Hospital and Associate Chairman, Department of Pediatric Oncology at Dana-Farber Cancer Institute. He is Director of the Pediatric Hematology/Oncology Fellowship Training Program at BCH/DFCI. He was a Howard Hughes Medical Institute Investigator for 16 years and his laboratory has been continuously NIH funded since 1986. He has trained over 45 fellows and post-doctoral fellows and numerous residents and medical students in my laboratory, the majority of which are still in academic medicine. He is a member of the Institute of Medicine of the National Academy of Sciences. He has published over 250 peer-reviewed manuscripts and textbook chapters. He formerly served on the NIH Recombinant DNA Advisory Committee and Gene Therapy Safety Assessment Board. He is actively involved in gene therapy trials for immunodeficiency and neurological genetic diseases and has been the investigator, co-investigator or sponsor (IND holder) of four previous gene therapy trials and is sponsor or investigator of three current trials. He has recently served as a councilor for the American Society of Hematology (ASH) and served on the Joint Oversight Committee of ASH/European Hematology Association Translational Research Training in Hematology. He is currently Vice-President (President-elect) of ASH. He served as the Editor-In-Chief of *Molecular Therapy* from 2004-2009. He is co-founder of the Transatlantic Gene Therapy Consortium. His basic research has focused on hematopoietic stem cell biology, including genetic diseases of the blood and specifically molecular and biochemical analysis of the interaction between hematopoietic stem cells and the bone marrow supporting environment.

ANNE WILLIS, M.A., George Washington University Cancer Institute

A long-term Ewing's Sarcoma survivor, Anne Willis, MA, is the Director of the Division of Cancer Survivorship at the George Washington University Cancer Institute (GWCI) and Director of the GW Center for the Advancement of Cancer Survivorship, Navigation and Policy (caSNP). She oversees the institute's local and national survivorship initiatives, including education programs for cancer survivors, capacity building for cancer centers and cancer survivorship research. Ms. Willis also directs caSNP efforts, including education and training programs for health care professionals, meetings on navigation and survivorship and health policy initiatives. Previous to GWCI, Ms. Willis was the Director of Survivorship Programs for the National Coalition for Cancer Survivorship where she developed and disseminated evidence-based programs, including the award-winning Cancer Survival Toolbox®, to empower people with cancer to advocate for themselves across the survivorship continuum.

LYNNE YAO, M.D., Center for Drug Evaluation and Research, FDA

Lynne Yao, MD, is the Associate Director, Office of New Drugs, Pediatric and Maternal Health Staff. The Pediatrics and Maternal Health Staff works within CDER and across FDA Centers to promote and necessitate the study of drug and biological products in the pediatric population and improve pregnancy and lactation-related information in product labeling. Previously, she was a Medical Officer Team Leader in the Division of Gastroenterology and Inborn Errors Products (2009-2012). The Inborn Errors of Metabolism Team is a review team that focuses on products for rare, inherited metabolic diseases including many biological products used as enzyme replacement therapies for these disorders. Dr. Yao is board-certified in Pediatrics and Pediatric Nephrology.

ANNE ZAJICEK, M.D., Pharm.D., The *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, NIH

Anne Zajicek, M.D., Pharm.D., is a board-certified pediatrician and pediatric clinical pharmacologist who currently serves as Chief of the Obstetric and Pediatric Pharmacology and Therapeutics Branch at the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD). She received a Bachelor's degree in Pharmacy from Duquesne University and a PharmD from the State University of New York at Buffalo; completed a postdoctoral fellowship in the Department of Pharmaceutics of St. Jude Children's Research Hospital; and served as an assistant professor at the University of Colorado School of Pharmacy and a Clinical Pharmacist at National Jewish Hospital and Research Center. In 1991, Dr. Zajicek entered medical school at the University of Pittsburgh, and, in 1998, completed a residency in pediatrics at the Children's Hospital of Pittsburgh. She practiced primary care pediatrics for 2 years and then continued her training as a pediatric clinical pharmacology fellow at Stanford University. She subsequently joined the U.S. Food and Drug Administration (FDA) in the Office of Clinical Pharmacology and Biopharmaceutics. She joined NICHD as a Pediatric Medical Officer in August 2003, and was appointed Chief of the Obstetric and Pediatric Pharmacology

and Therapeutics Branch in 2010. The Branch is responsible for the NIH implementation of the Best Pharmaceuticals for Children Act, and manages a portfolio of basic, translational and clinical research and training grants in obstetric and pediatric pharmacology.